PDGFB gene

platelet derived growth factor subunit B

Normal Function

The *PDGFB* gene provides instructions for making one version (isoform) of the platelet derived growth factor (PDGF) protein. This protein is involved in many cellular processes, including cell growth and division (proliferation), maturation (differentiation), and movement. The *PDGFB* gene provides instructions for a precursor protein that must be processed to be able to perform its function. Before processing, the precursor PDGFB protein attaches (binds) to another PDGFB protein or a similar protein called the PDGFA precursor protein, forming a structure known as a dimer. Once the dimer is formed, the precursor proteins are processed by being cut at specific locations, which forms the functional (active) PDGF proteins, called PDGF-BB and PDGF-AB.

The active PDGF-BB or PDGF-AB protein binds to a PDGF receptor, which initiates cellular signaling. PDGF signaling activates many pathways important in cell proliferation, differentiation, and movement.

Health Conditions Related to Genetic Changes

dermatofibrosarcoma protuberans

Dermatofibrosarcoma protuberans, a rare type of cancer that causes a tumor in the deep layers of skin, is characterized by a somatic mutation involving the *PDGFB* gene. Somatic mutations are not inherited, but are acquired during a person's lifetime and are present only in certain cells. Dermatofibrosarcoma protuberans is associated with a rearrangement (translocation) of genetic material between chromosomes 17 and 22. This translocation, written as t(17;22), fuses part of the *PDGFB* gene on chromosome 22 with part of another gene on chromosome 17 called *COL1A1*. The translocation is found on one or more extra chromosomes that can be either the normal linear shape or circular. The resulting combined (fusion) gene is called *COL1A1-PDGFB*.

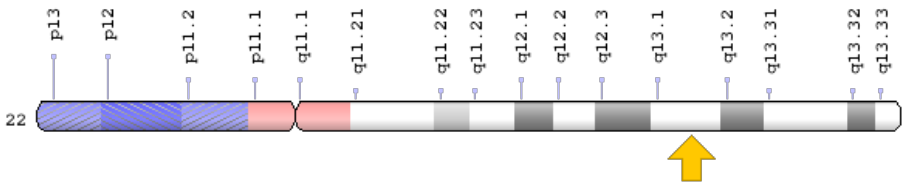
The *COL1A1-PDGFB* fusion gene provides instructions for making a fusion protein. In the translocation, the *PDGFB* gene loses the part of its DNA that inhibits its activity, and production of the COL1A1-PDGFB fusion protein is controlled by *COL1A1* gene sequences. As a result, the gene fusion leads to the production of large amounts of the fusion protein. The COL1A1-PDGFB protein forms a dimer and is processed like the normal PDGFB precursor protein. Processing removes the COL1A1 portion, which forms a protein that researchers believe functions like the active PDGF-BB protein. Excess PDGF-BB protein abnormally

stimulates cells to proliferate and differentiate, leading to the tumor formation seen in dermatofibrosarcoma protuberans.

Chromosomal Location

Cytogenetic Location: 22q13.1, which is the long (q) arm of chromosome 22 at position 13.1

Molecular Location: base pairs 39,223,359 to 39,245,055 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- becaplermin
- c-sis
- FLJ12858
- PDGF-2
- PDGF subunit B
- PDGF, B chain
- PDGF2
- platelet-derived growth factor 2
- platelet-derived growth factor beta polypeptide
- platelet-derived growth factor subunit B
- platelet-derived growth factor, B chain
- SIS
- SSV

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PDGFB%5BTIAB%5D%29+OR+%28platelet-derived+growth+factor+beta+polypeptide%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- PLATELET-DERIVED GROWTH FACTOR, BETA POLYPEPTIDE
  http://omim.org/entry/190040

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/PDGFBID155.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PDGFB%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8800
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5155
- UniProt
  http://www.uniprot.org/uniprot/P01127

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